

METHODS FOR DIAGNOSIS AND TREATMENT OF BLOOM'S SYNDROME

Abstract of the Disclosure

5 The present invention provides a method for  
diagnosing BS as well as determining whether a subject is a  
carrier of a mutated *BLM* gene. The present invention also  
provides one or more single-stranded nucleic acid probes and  
antibodies which may be formulated in kits, and used for  
diagnosing BS or determining whether a subject is a carrier  
of a mutated *BLM* gene. In addition, the present invention  
10 provides a method for treating or preventing the onset of BS  
in a subject in need of such treatment or prevention, as well  
as vectors and stem cells useful for such treatment or  
prevention. The present invention also provides a purified  
and isolated nucleic acid encoding an enzymatically active  
15 *BLM* protein, a vector comprising this nucleic acid, a cell  
stably transformed with this vector, as well as a method for  
producing recombinant, enzymatically active *BLM* protein. A  
purified, enzymatically active *BLM* protein is also provided  
by the present invention. Finally, the present invention  
20 provides a vector, an embryonic stem cell, and a non-human,  
transgenic animal, each of which comprises a mutated *BLM*  
gene, as well as a method for producing the non-human,  
transgenic animal.